

ELASTIN DISORDERS

- * CUTIS LAXA
↓ COPPER
[AUTOSOMAL RECESSIVE → MORE SEVERE
AUTOSOMAL DOMINANT
X-LINKED CALLED OCCIPITAL HORN SYNDROME → THIS FORM OF DISORDER IS CONSIDERED A MILD TYPE
MENKES SYNDROME
- * SUPRAVALVULAR AORTIC STENOSIS (SVAS) → **PRESENT FROM BIRTH THAT CHARACTERIZED BY
A NARROWING OF LARGE BLOOD VESSEL
DOMINANT
- * WILLIAMS SYNDROME → * DELETION (STRUCTURAL ABERATION)
DOMINANT
- * MARFAN SYNDROME → CAN CAUSE DEFECTS OF THE HEART VALVES & AORTA
DOMINANT

MELANIN DISORDERS

- * ALBINISM → OCULOCUTANEOUS ALBINISM → MOST SEVERE FORM
• ALBINISM CAN BE RECESSIVE & CAN BE DOMINANT
BUT IN MOST CASES **RECESSIVE**
- * HYPERPIGMENTATION → DARKENING OF AN AREA OF SKIN OR NAILS CAUSED BY ↑↑ MELANIN
VERY COMMON & HARMLESS